Advisory Committee of Heritable Disorders and Genetic Diseases in Newborns and Children

R. Rodney Howell, M.D.
Chair
ACHDGDNC
February 28, 2005

Current Environment, With Emphasis on Newborn Genetic Testing

- Changing technology
  - Multiplex testing platforms rapidly developing
- Large numbers of individually rare diseases
  - Few providers with required expertise
- New technology on the horizon

Current Environment, continued

- Legislation for Heritable Disorders Program
  - Advisory Committee on Heritable Disorders and Genetic Diseases in Newborns and Children
  - Grant program: HRSA's Regional Collaboratives

- Recommendations from the ACMG-Newborn Screening Expert Group
  - Extensive report, developed under a contract from HRSA now complete after more than two years' work by a large and diverse group of persons

Legislation

- **Title XXVI of the Children's Health Care Act of 2000 (Title XI of PHS act)**
  - **Establishes the Heritable Disorders Program to improve the ability of States to provide newborn and child screening for heritable disorders.**
  - **Three Sections in legislation:**
    - **PHS Act: Section 1109**
    - **PHS Act: Section 1110**
    - **PHS Act: Section 1111- Advisory Committee on Heritable Disorders**

Legislation

Section 1109 provides funds for grants to establish or expand or improve:

- **Systems or Programs:**
  - **To provide screening, counseling or health care services to newborns and children having or at risk for heritable disorders**

Section 1110 authorizes the Secretary to award grants to:

- **Provide for demonstration programs**
  - **To evaluate the effectiveness of screening, counseling or health care services in reducing the morbidity and mortality caused by heritable disorders in newborns and children.**

Advisory Committee on Heritable Disorders and Genetic Diseases in Newborns and Children
PHS Act: Section 1111

PURPOSE OF THE COMMITTEE

- **Provide the Secretary advice and recommendations concerning the grants and projects authorized under section 1109 and technical information to develop policies and priorities for this program that will enhance the ability of State and local health agencies to provide for newborn and child screening, counseling and health services for newborns and children having or at risk for heritable disorders.**

PURPOSE OF THE COMMITTEE

- **Specifically, the committee shall advise and guide the Secretary regarding the most appropriate application of universal newborn screening tests, technologies, policies, guidelines and programs for effectively reducing morbidity and mortality in newborns and children having or at risk for heritable disorders.**

Advisory Committee on Heritable Disorders

- Medical, technical, or scientific professionals with special expertise in heritable disorders, or in providing screening, counseling, testing or specialty services for newborns and children at risk for heritable disorders;
- Members of the public having special expertise about or concern with heritable disorders; and
- Representatives from such Federal agencies, public health constituencies, and medical professional societies as determined to be necessary by the Secretary, to fulfill the duties of the Advisory Committee

Committee Members

William J. Becker, D.O., M.P.H.
Bureau of Public Health Laboratories
Ohio Department of Health

Amy Brower, Ph.D.
Third Wave Technologies

Peter B. Coggins, Ph.D.
PerkinElmer Life and Analytical Science

Committee Members

E. Stephen Edwards, M.D., F.A.A.P.
American Academy of Pediatrics
Past President

Gregory A. Hawkins, Ph.D.
Department of Internal Medicine
Wake Forest University School of Medicine

Committee Members

Jennifer L. Howse, Ph.D.
March of Dimes Birth Defects Foundation

R. Rodney Howell, M.D.
(Chairperson)
The University of Miami School of Medicine
Department of Pediatrics

Committee Members

Piero Rinaldo, M.D., Ph.D.
Mayo Clinic

Derek Robertson, M.B.A., J.D.
Parent

<u>Ex-officio Members (voting)</u>
Peter C. van Dyck, M.D., M.P.H.
Health Resources and Services Administration
- HRSA manages Committee

Committee Members

Ex-officio Members, continued

Denise Dougherty, Ph.D.
Agency for Healthcare Research and Quality

Coleen Boyle, Ph.D.
Centers for Disease Control and Prevention

Duane Alexander, M.D.
National Institutes of Health

Committee Members

<u>Liaison Members (non-voting)</u>

James W. Collins, Jr., M.D., M.P.H.
Advisory Committee on Infant Mortality

Joseph Telfair, Dr. P. H., M.S.W.- M.P.H.
Advisory Committee on Genetics, Health and Society

This report contains the collective views of an international group of experts and does not necessarily represent the decisions or the stated policy of the World Health Organization.

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TECHNICAL REPORT SERIES

No. 401

SCREENING FOR INBORN ERRORS OF METABOLISM

Report of a WHO Scientific Group

WORLD HEALTH ORGANIZATION

Newborn Screening for Genetic Disease

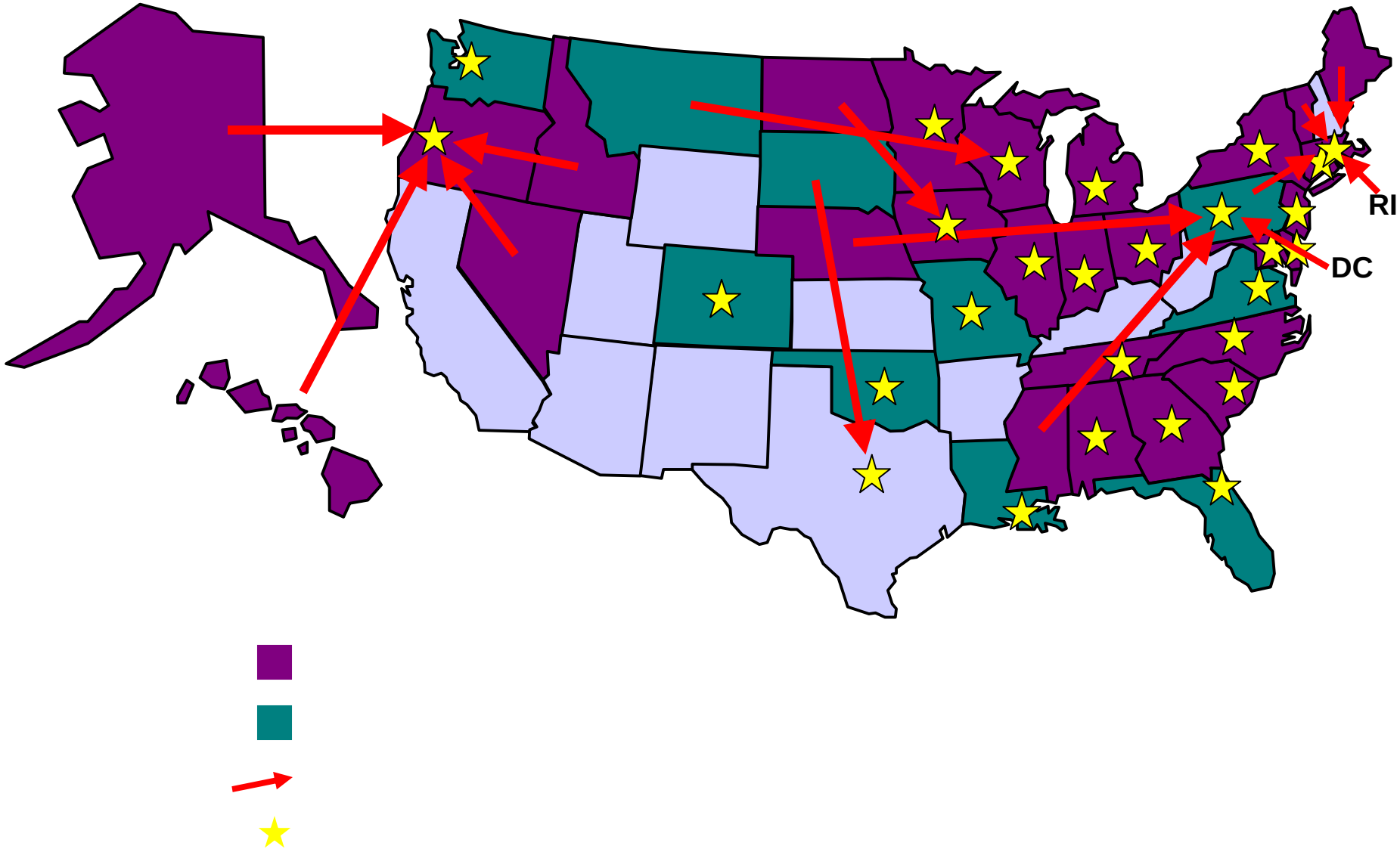
- In March of 2003, the General Accounting Office reported to Congress on
- Newborn Screening: The characteristics of the State Programs
- Over 4,000,000 infants are screened annually in this country, making newborn screening by far the most common type of genetic testing done today
- Most states have similar programs for funding and administering their programs, funded largely through fees from health care providers

Newborn Screening is a State Public Health Program

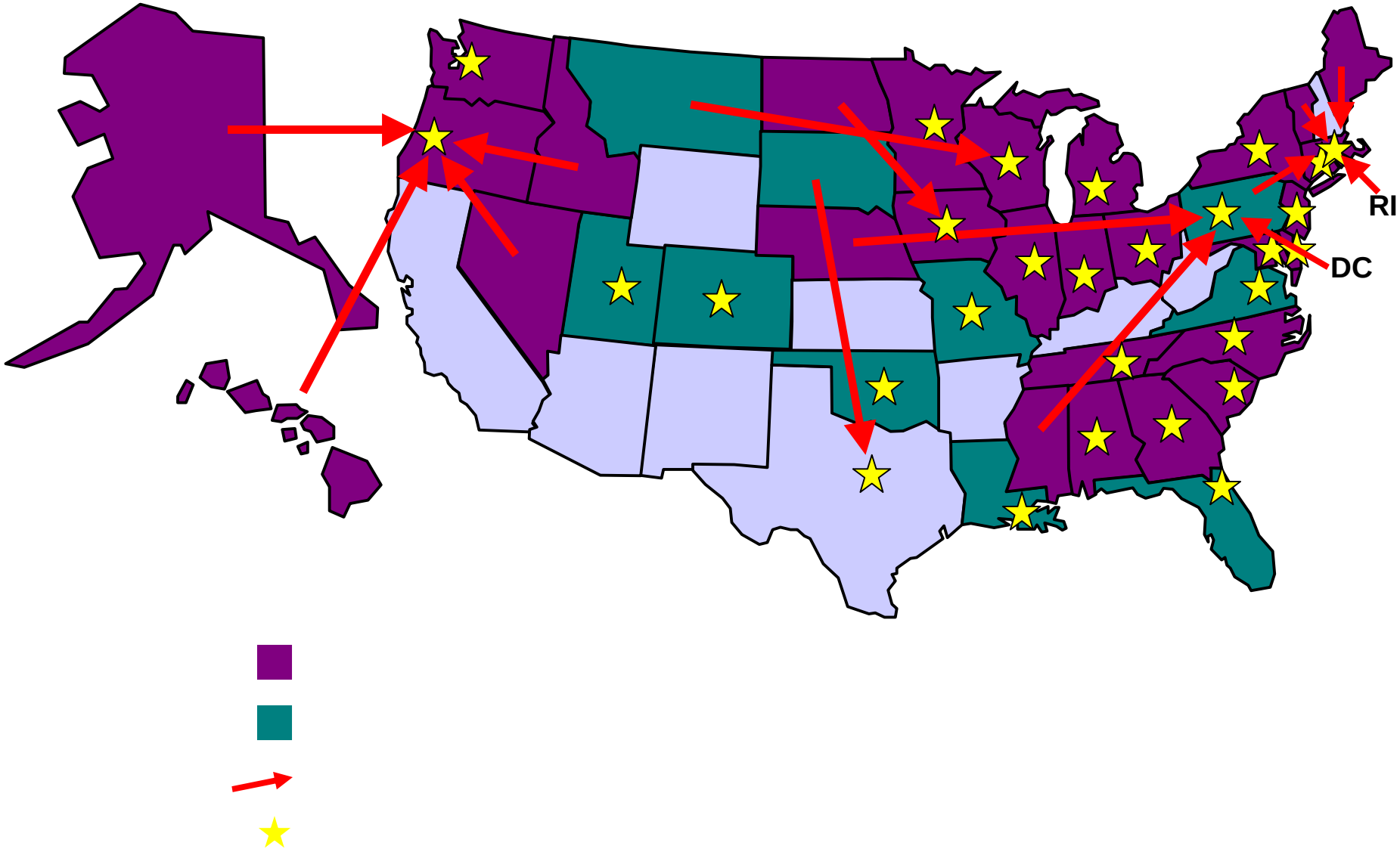
- Routine newborn screening has been carried out in all 50 states since the 1970s
- Conditions such as phenylketonuria, with simple reliable screening tests and proven treatment efficacy has been the targets of testing
- Congenital hypothyroidism and a handful of other diseases have been added on a state by state basis
- There is extraordinary variation from state to state and there has been little systematic evaluation of either the rational for screening and/or the outcomes of such screening

New Technology Has Permitted The Rapid Expansion of Newborn Screening

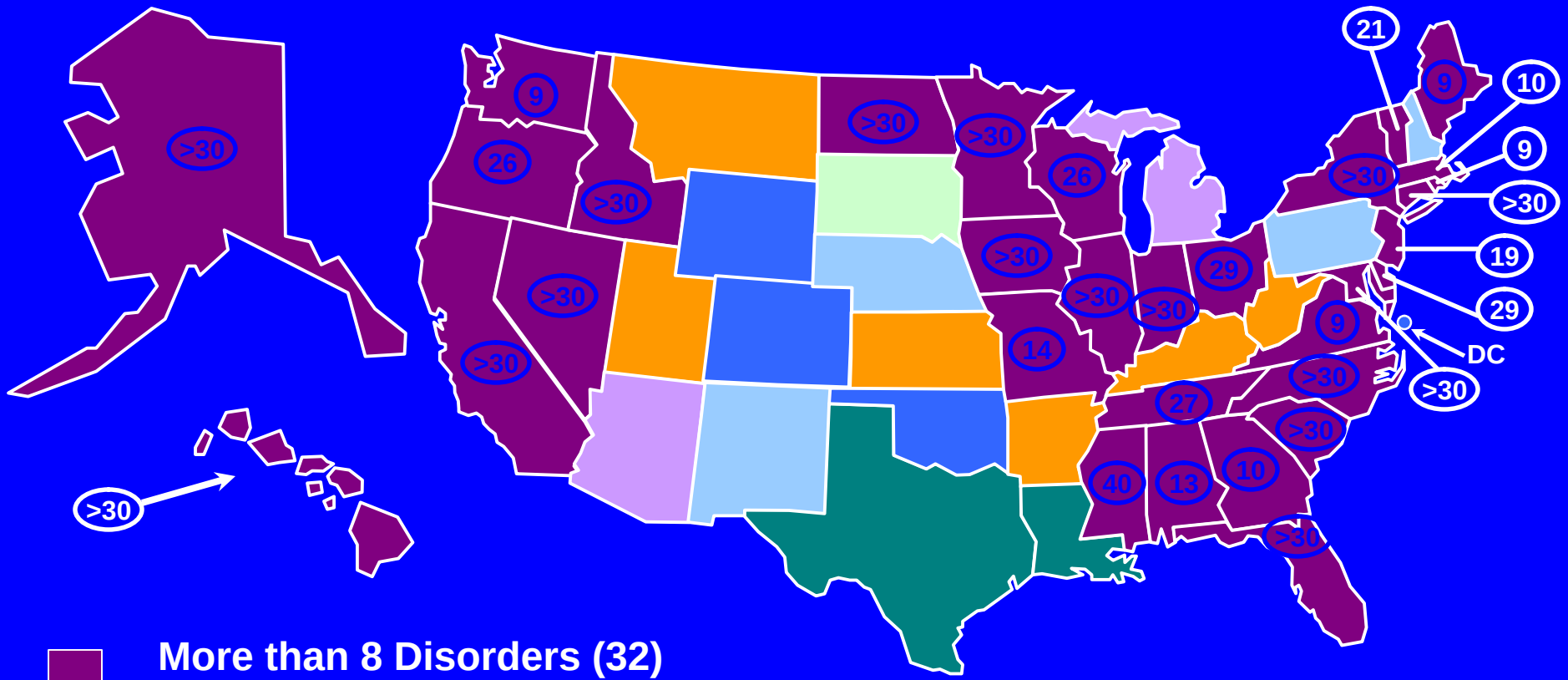
- Historically, newborn screening for conditions such as phenylketonuria has required an individual, specific test for each disorder
- The widespread use of tandem mass spectroscopy has permitted the identification of a large number of analytes important in the diagnosis of diseases in the newborn using a single technology



States With MS/MS Screening
September 2004



States With MS/MS Screening
January 2005



More than 8 Disorders (32)

[More than 30 Disorders (14)]

8 Disorders (2)

7 Disorders (4)

6 Disorders (4)

5 Disorders (2)

4 Disorders (6)

3 Disorders (1)

U.S. Newborn Screening

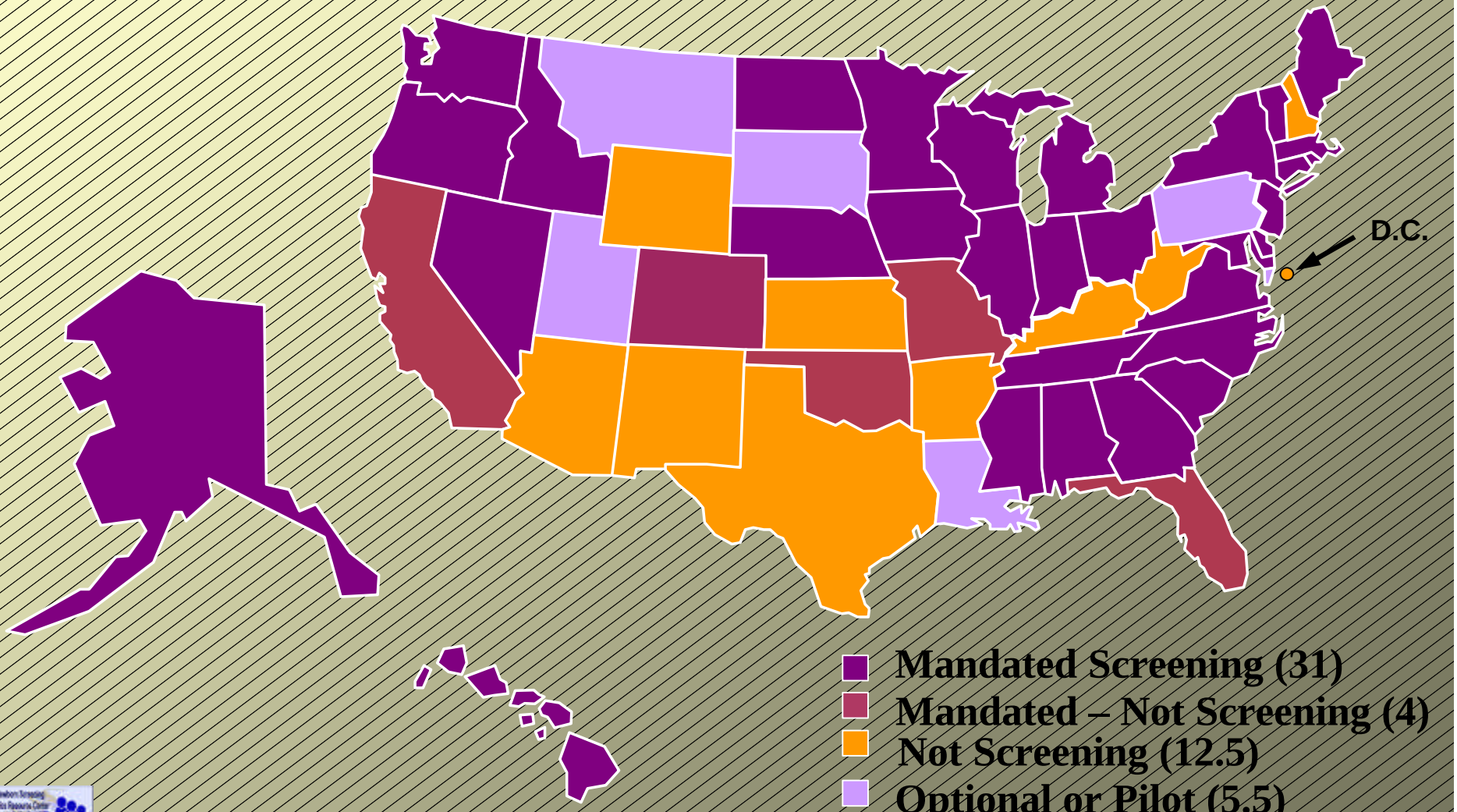
Mandated Disorders – Jan 2005

(Note: Other disorders may be offered but are not mandated)

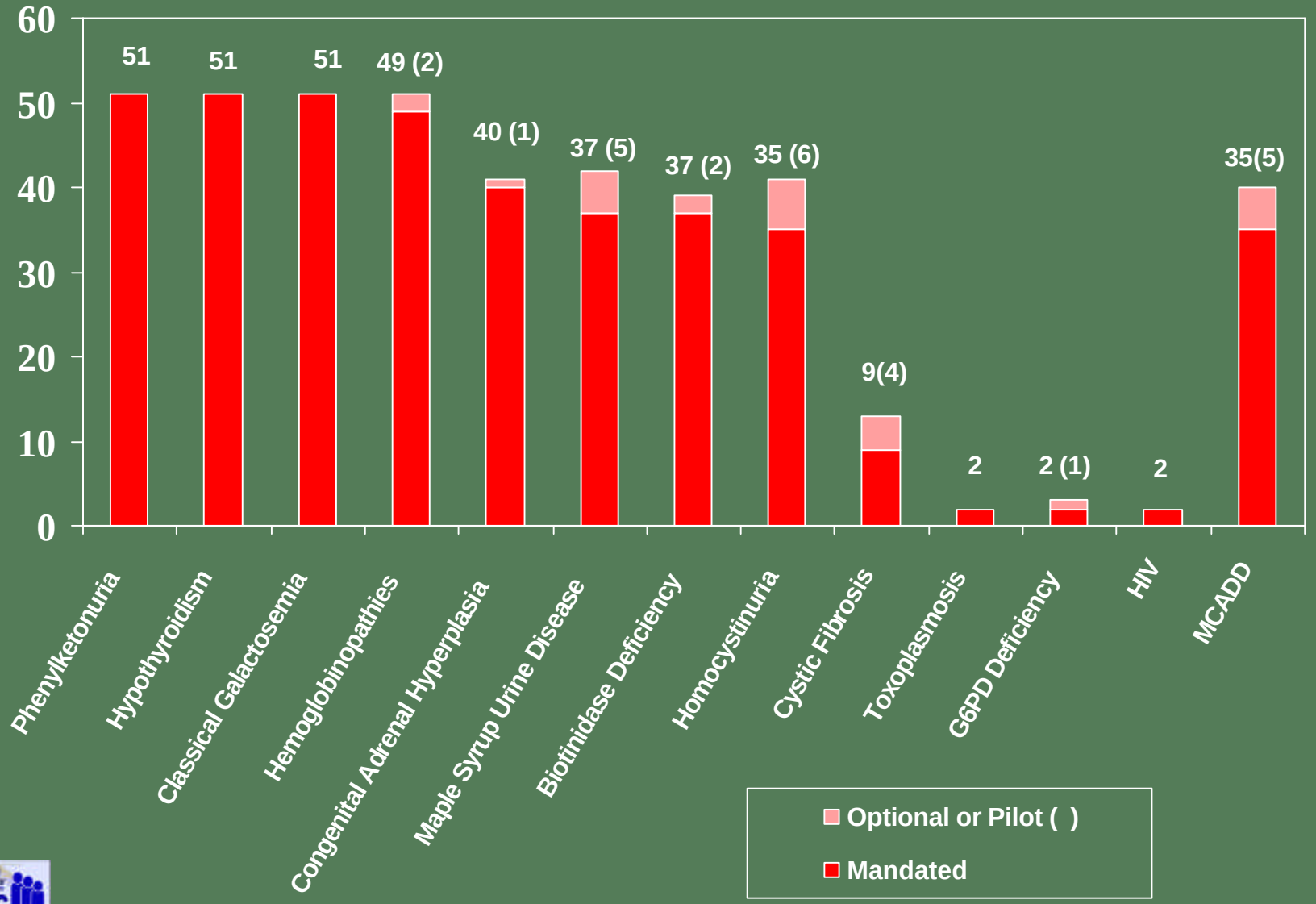
NNSGRC

U.S. Newborn Screening – 2005

Medium Chain Acyl-CoA Dehydrogenase Deficiency



Disorders Screened in United States January 2005



NNSGRC

Recent Screening Changes

- Alabama → + BIO + 6 MS/MS Conditions
- Connecticut → + MS/MS beyond 3
- Florida → + CF + BIO + MS/MS Conditions
- Georgia → Began MCAD (previously mandated)
- Louisiana → Began pilot for 5 MS/MS Conditions
- Michigan → + 3 MS/MS
- Minnesota → + BIO
- New York → + MS/MS beyond MCAD
- North Carolina → + BIO
- South Carolina → + CF + BIO + 20 MS/MS beyond amino acids

Advisory Committee on Heritable Disorders and Genetic Diseases in Newborns and Children

- The Committee has held three meetings which have focused on the current status of newborn screening and related systems
- The next meeting is scheduled for April 21-22, 2005 at the Ronald Reagan Building and International Trade Center, Pennsylvania Avenue in Washington, D. C.
- Public attendance and comment are invited. Public Comment must be scheduled with Executive Secretary, shown on my final slide

Issues Identified

- **Recommendations for uniform panel of conditions based on the panel prepared by experts and presented to Advisory Committee**

- **Assessing capacity needs of states/newborn screening programs: Three subcommittees have been formed:**
  - **Education and Training**
    - **Parent Education/notification/informed decision-making**
    - **Workforce needs**
  - **Follow-up and Treatment**
  - **Laboratory Standards and Procedures**

Advisory Committee on Heritable Disorders and Genetic Diseases in Newborns and Children

mchb.hrsa.gov/programs/genetics/committee/

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